DOCUMENT RESUME

ED 425 553 EC 306 214

TITLE	Prader-Willi Syndrome. ARC Q&A #101-52.
INSTITUTION	Arc, Arlington, TX.
SPONS AGENCY	Department of Energy, Washington, DC.
PUB DATE	1997-03-00
NOTE	4p.
CONTRACT	DE-FG03-96ER62162
AVAILABLE FROM	Arc National Headquarters, 500 E. Border St., S-300, Arlington, TX 76010; Tel:: 817-261-6003; TDD: 817-277-0553.
PUB TYPE	Guides - Non-Classroom (055)
EDRS PRICE	EDRS Price MF01 Plus Postage. PC Not Available from EDRS.
DESCRIPTORS	Behavior Problems; Clinical Diagnosis; Congenital Impairments; Eating Disorders; Genetics; Incidence; *Intervention; *Mental Retardation; Neurology; Obesity; *Special Health Problems; *Symptoms (Individual Disorders); Therapy
IDENTIFIERS	*Prader Willi Syndrome

ABSTRACT

This fact sheet uses a question-and-answer format to summarize what is known about Prader-Willi Syndrome (PWS), a complex genetic disorder resulting in short stature, mental retardation or learning disabilities, incomplete sexual development, characteristic behavior problems, low muscle tone, and an involuntary urge to eat constantly, which results in obesity. Questions and answers address the following topics: what Prader-Willi Syndrome is; the inheritability of PWS; incidence of PWS (about 1 in 14,000); infants with PWS and development of the typical eating disorders; the defect in the hypothalamus which causes the overeating; ways to control the PWS individual's eating; other characteristics of PWS; the genetics of PWS; typical behavior problems of people with PWS; types of intervention and care which allow a reasonably normal life; the importance of early diagnosis; how to determine if one's child has PWS; and sources of more information. (DB)

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Prader-Willi Syndrome

ARC Q&A #101-52

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Q&A

Prader-Willi Syndrome

by Prader-Willi Syndrome Association (USA)

What is Prader-Willi syndrome (PWS)?

PWS is a complex genetic disorder that includes short stature, mental retardation or learning disabilities, incomplete sexual development, characteristic behavior problems, low muscle tone, and an involuntary urge to eat constantly, which, coupled with a reduced need for calories, leads to obesity.

Does PWS run in families?

Although PWS is associated with an abnormality of chromosome 15, it is generally considered not to be an inherited condition, but rather a spontaneous genetic birth defect that occurs at or near the time of conception. PWS is found in people of both sexes and all races.

How common is this problem?

About 1 in 14,000 people in the U.S. are estimated to have PWS, and the birth rate may be even higher. Prader-Willi syndrome is one of the 10 most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified.

Does the eating problem associated with PWS begin at birth?

No, newborns with PWS are typically described as "floppy" and are unable to suck well enough to get sufficient nutrients -- due to the low muscle tone (hypotonia). Often they must be fed through a tube for several months after birth, until muscle control improves. Sometime in the following years, usually by preschool age, children with PWS develop an increased interest in food and quickly gain excessive weight if calories are not restricted.

Why do people who have PWS eat so much?

People with PWS have a flaw in the part of their brain (the hypothalamus) that determines hunger and satiety (fullness). These people never feel full enough, so they have a continuous urge to eat. To compound this problem, people with PWS need considerably fewer calories than normal to maintain an appropriate weight. The obesity that results is the major cause of illness and death in this disorder. As in the general population, obesity in PWS can cause high blood pressure, respiratory difficulties, diabetes and other problems.

Can anything be done to control the eating?

Unfortunately, no appetite suppressant has worked consistently for people with PWS. Most must be on an extremely low-calorie diet all their lives and must have their environment designed so that they have very limited access to food. For example, many families have to lock the kitchen or the cabinets and refrigerator.

I know many people who eat a lot and have obesity. How do I know they don't have PWS?

There's more to PWS than the obesity. People with PWS have a characteristic appearance and speech quality, significant learning disabilities or mental retardation, and various other problems. A number of these features must be present for a clinical diagnosis of PWS, and specific genetic tests are available to confirm the diagnosis.

What is known about the genetic abnormality?

Basically, the occurrence of PWS is due to the absence of a few genes on one of the chromosomes that must affect functioning of the hypothalamus, among other things. This is an area of very active research in a number of laboratories around the world, since understanding this defect may be very helpful not only to people with PWS but also to understanding obesity, mental retardation, and behavior in otherwise normal people.

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What kinds of behavior problems do people with PWS have?

In addition to sometimes extreme attempts to obtain food, people with PWS are prone to temper outbursts, stubbornness, rigidity, argumentativeness, and repetitive thoughts and behaviors. Strategies to deal with these problems usually include structuring the person's environment, implementing behavioral management techniques, and occasionally drug therapy.

Is it possible for people with PWS to lead normal lives?

People with PWS can accomplish many of the things their "normal" peers do -- attend school, enjoy community activities, get jobs, and even move away from home. However, they need a lot of help. School children with PWS are likely to need special education and related services, such as speech and occupational therapy. In community, work and residential settings, adolescents and adults often need special assistance to learn and carry out responsibilities and to get along with others. In all settings, people with PWS need around-the-clock food supervision. As adults, most affected individuals do best in a special group home for people with PWS, where food access can be restricted without interfering with those who do not need such restriction. Although in the past many died in adolescence or young adulthood, it is thought that prevention of obesity will allow a person with PWS to live a normal lifespan.

Does early diagnosis help?

Early diagnosis of Prader-Willi syndrome gives parents an opportunity to manage their child's diet and avoid obesity and its related problems from the start. Since infants and young children with PWS typically have developmental delays in all areas, diagnosis may facilitate a family's access to critical early intervention services and help identify areas of need or risk. Diagnosis also makes it possible for families to get information and support from professionals and other families who are dealing with the syndrome.

If someone believes that they or their child may have PWS, how do they find out?

The best way would be to contact their health care provider and ask to be evaluated for Prader-Willi syndrome. Formal diagnostic criteria for the clinical recognition of PWS have been published (Holm et al, 1993). Many doctors will choose to refer to a medical geneticist who specializes in diagnosing and testing for genetic conditions such as PWS. After taking a history and doing a physical examination, the diagnostician will arrange for specialized genetic testing to be done on a blood sample to evaluate for the genetic abnormality found in people with PWS.

How can a person get more information about PWS?

There is a strong national organization of families and professionals dealing with PWS called the Prader-Willi Syndrome Association (USA). It has a toll-free number (1-800-926-4797) and is organized to answer questions and help individuals deal with the problems associated with PWS. PWSA (USA) provides a newsletter and other publications, an annual national conference, and chapters throughout the country to provide family support and advocacy. One also could call a local genetics unit to ask for information on PWS.

Reference

Holm, V., et al. (1993). *Prader-Willi Syndrome; Consensus Diagnostic Criteria.* Pediatrics, 91, 398-402.

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Funded by the U.S. Department of Energy ELSI Program under Grant No. DE-FG03-96ER62162. Support from DOE does not constitute an endorsement of the views expressed in this publication.

#101-52 **March 1997**